Sponsored by;

Macedonian Academy of Arts and Sciences (MASA), Skopje

4TH RARE DISEASE SOUTH EASTERN EUROPE (SEE) MEETING SKOPJE, MACEDONIA

(EAP, UEMS Section of Paediatrics)

Organizing Committee:

Momir Polenakovic, President
Zoran Gucev, Secretary
Vladimir Serafimoski
Nada Pop-Jordanova
Ilija Filipche
Zivko Popov
Velibor Tasic
Aspazija Sofijanova
Vesna Aleksovska
Dijana Plaseska Karanfilska

SATURDAY, NOVEMBER 14

Rare Diseases in SEE

Moderators: Nada Pop-Jordanova, Zoran Gucev

09.00-09.30 **Welcome and opening**

Mrs. Maja Ivanova, First Lady of the Republic of Macedonia, Honorary Patron of the Association of citizens for rare diseases "Life with Challenges"

Mr. Vlado Kambovski, President, MASA

Mr. Nikola Todorov, Minister of Health, Macedonia

Mr. Saso Stefanoski, Director, Health Fund of Macedonia

Mr. Momir Polenakovic, Director, Research Center for Genetic Engineering and Biotechnology "G.D.Efremov", MASA

Mrs. Aspazija Sofijanova, Director, University Pediatric Clinic, Skopje, Macedonia

Mrs. Vesna Aleksovska, President of "Life With Challenges", Skopje, Macedonia

Session I

09.30–09.50 **Momir Polenakovic, Velibor Tasic, Zoran Gucev, Skopje, Macedonia**
Rare diseases: an ongoing battle to curb the costs of diagnosis and treatment

09.50-10.10 **Velibor Tasic, Skopje, Macedonia**
Steroid resistant nephrotic syndrome

10.10-10.30 **Hadil Kathom, Sofia, Bulgaria**
Phenilketonuria

10.30-10.50 **Stayko Sarafov, Sofia Bulgaria**
Epidemiology, clinical features, differential diagnosis and treatment of TTR-FAP in Bulgaria

10.50-11.10 **Ivailo Tournev, Sofia Bulgaria**
Selective screening, carriers testing and carriers follow-up program of the Bulgarian Neuromuscular Disease Society

11.10-11.30 **Vukasin Andric, Zagreb, Croatia**
Pompe disease – recognizing and enzyme replacement therapy

Discussion

11.30-11.50

11.50-12.10 **Coffee break**

12.10-12.30 **Shire symposia**

Gunter Harms, Berlin, Germany
Why are orphan drugs different? Achieving sustainable access to treatment for rare disease patients

12.30-12.50 **Gudrun Schlegel, Hamburg-Eppendorf, Germany**
Mental retardation in PKU: The role of microglia

12.50-13.10 **Stéphane Demotz, Lausanne Switzerland**
GM1-gangliosidosis and Morquio disease type B

13.10-13.30 **Eduard Pashke, Graz, Austria**
Recent developments in the laboratory diagnosis of MPS diseases

13.30-13.50 **Zoran Gucev, Skopje, Macedonia**
The Proteus Spectrum

13.50-14.10 **Velibor Tasic, Skopje, Macedonia**
Idiopathic infantile hypercalcemia

14.10-14.30 **Venko Filipce, Skopje, Macedonia**
Selective and superselective angiography of pediatric moyamoya disease angioarchitecture in the anterior and posterior circulation

14.30-14.50 **Arben Taravari, Skopje, Macedonia**
Huntington's horea, etiopatogenesis, clinical manifestations and contemporary therapeutical approach

Discussion

Conclusion remarks

Zoran Gucev, Velibor Tasic, Momir Polenakovic

Venue

Macedonian Academy of Arts and Sciences
Bul. Krste Misirkov, No.2, Skopje
Republic of Macedonia

Language of the Meeting: English

Certificates of Attendance will be provided by Prof. V. Tasic